

## WHAT IS CLAIMED IS:

1. A method for detecting a DNA having the mitochondrial DNA 3243 mutation, comprising performing PCR using a DNA obtained from a sample as a template and detecting an amplification product, wherein primers used for PCR comprise a primer having a nucleotide sequence complementary to a nucleotide sequence starting from the nucleotide number 243 in the nucleotide sequence of SEQ ID NO: 1 and having a length of 12 to 30 nucleotides.

2. The method according to claim 1, wherein the primers used for PCR comprises a primer having the nucleotide sequence of SEQ ID NO: 3 and a primer having the nucleotide sequence of SEQ ID NO: 5.

3. A method for quantifying a DNA having the mitochondrial DNA 3243 mutation, comprising performing quantitative PCR using a DNA obtained from a sample as a template and quantifying an amplification product, wherein the quantitative PCR is a method of using a system in which fluorescence changes depending on amount of an amplification product to quantify the amplification product in PCR on real time basis by measurement of fluorescence and quantifying the DNA in the sample on the basis of the result of the measurement, and primers used for the quantitative PCR comprise a primer having a nucleotide sequence complementary to a nucleotide sequence starting from the nucleotide number 243 in the nucleotide sequence of SEQ ID NO: 1 and having a length of 12 to 30 nucleotides.

4. The method according to claim 3, wherein the primers used for the quantitative PCR comprises a primer having the nucleotide sequence of SEQ ID NO: 3 and a primer having the nucleotide sequence of SEQ ID NO: 5.

5. A method for determining the heteroplasmy ratio of the mitochondrial DNA 3243 mutation contained in a

sample, which comprises:

- (a) quantifying a DNA having the mitochondrial DNA 3243 mutation by the method as defined in claim 3 or 4,
- (b) quantifying mitochondrial DNAs by a method for quantifying mitochondrial DNAs comprising performing quantitative PCR using a DNA obtained from a sample as a template and quantifying an amplification product, wherein the quantitative PCR uses a system in which fluorescence changes depending on amount of an amplification product to quantify the amplification product in PCR on real time basis by measurement of fluorescence, and DNAs in the sample is quantified on the basis of the result of the measurement, and
- (c) calculating the heteroplasmy ratio of the mitochondrial DNA 3243 mutation from the results of (a) and (b).

6. The method according to claim 5, wherein primers used in the step of (b) comprise a primer having the nucleotide sequence of SEQ ID NO: 3 and a primer having the nucleotide sequence of SEQ ID NO: 4.

7. The method according to any one of claims 3 to 6, wherein the system comprises a nucleic acid probe of which 5' end is labeled with a fluorescent dye, and in which fluorescence of the fluorescent dye decreases upon hybridization, wherein the nucleic acid probe has a nucleotide sequence starting from the nucleotide number 212 or 215 in the nucleotide sequence of SEQ ID NO: 1 and having a length of 15 to 40 nucleotides or a nucleotide sequence starting from the nucleotide number 222 in the nucleotide sequence of SEQ ID NO: 2 and having a length of 15 to 40 nucleotides, and fluorescence of the fluorescent dye is measured.

8. A kit for the method as defined in claim 1, which comprises a primer having a nucleotide sequence complementary to a nucleotide sequence starting from the

nucleotide number 243 in the nucleotide sequence of SEQ ID NO: 1 and having a length of 12 to 30 nucleotides.

9. The kit according to claim 8, which comprises a primer having the nucleotide sequence of SEQ ID NO: 3 and a primer having the nucleotide sequence of SEQ ID NO: 5.

10. A kit for the method as defined in claim 3, which comprises a primer having a nucleotide sequence complementary to a nucleotide sequence starting from the nucleotide number 243 in the nucleotide sequence of SEQ ID NO: 1 and having a length of 12 to 30 nucleotides.

11. The kit according to claim 10, which comprises a primer having the nucleotide sequence of SEQ ID NO: 3 and a primer having the nucleotide sequence of SEQ ID NO: 5.

12. A kit for the method as defined in claim 5, which comprises a first primer pair comprising a primer having a nucleotide sequence complementary to a nucleotide sequence starting from the nucleotide number 243 in the nucleotide sequence of SEQ ID NO: 1 and having a length of 12 to 30 nucleotides, and a second primer pair for quantifying mitochondrial DNAs.

13. The kit according to claim 12, wherein the first primer pair comprises a primer having the nucleotide sequence of SEQ ID NO: 3 and a primer having the nucleotide sequence of SEQ ID NO: 5.

14. The kit according to claim 12, wherein the second primer pair comprises a primer having the nucleotide sequence of SEQ ID NO: 3 and a primer having the nucleotide sequence of SEQ ID NO: 4.

15. The kit according to any one of claims 10 to 14, which further comprises a nucleic acid probe of which 5' end is labeled with a fluorescent dye, in which fluorescence of the fluorescent dye decreases upon hybridization, and which has a nucleotide sequence starting from the nucleotide number 212 or 215 in the nucleotide sequence of SEQ ID NO: 1 and having a length of

15 to 40 nucleotides or a nucleotide sequence starting from the nucleotide number 222 in the nucleotide sequence of SEQ ID NO: 2 and having a length of 15 to 40 nucleotides.

16. A nucleic acid probe of which end is labeled with a fluorescent dye, and in which fluorescence of the fluorescent dye decreases upon hybridization, wherein the nucleic acid probe has a nucleotide sequence complementary to a nucleotide sequence starting from the nucleotide number 230 in the nucleotide sequence of SEQ ID NO: 2 and having a length of 14 to 40 nucleotides, and the 3' end of the probe is labeled with the fluorescent dye.

17. The nucleic acid probe according to claim 16, wherein the nucleic acid probe has the nucleotide sequence of SEQ ID NO: 21 or 22.

18. A method for detecting a mutation comprising performing a melting curve analysis for a nucleic acid having a single nucleotide polymorphism site by using a nucleic acid probe labeled with a fluorescent dye and measuring fluorescence of the fluorescent dye, and detecting the mutation on the basis of the result of the melting curve analysis, wherein the single nucleotide polymorphism is a mutation at the 3243rd position in a mitochondrial DNA, and the nucleic acid probe is the nucleic acid probe as defined in claim 16 or 17.

19. The method according to claim 18, wherein a region containing the single nucleotide polymorphism site in a nucleic acid contained in a sample is amplified to obtain the nucleic acid showing the single nucleotide polymorphism.

20. The method according to claim 19, wherein the amplification is performed by a method of using a DNA polymerase.

21. The method according to claim 20, wherein the amplification is performed in the presence of a nucleic

acid probe.

22. A kit for the method as defined in claim 18, which includes a nucleic acid probe of which end is labeled with a fluorescent dye, and in which fluorescence of the fluorescent dye decreases upon hybridization, wherein the nucleic acid probe has a nucleotide sequence complementary to a nucleotide sequence starting from the nucleotide number 230 in the nucleotide sequence of SEQ ID NO: 2 and having a length of 14 to 40 nucleotides, and the 3' end of the probe is labeled with the fluorescent dye.

23. The kit according to claim 22, wherein the nucleic acid probe has the nucleotide sequence of SEQ ID NO: 21 or 22.

24. The kit according to claim 22 or 23, which further comprises a primer for amplifying a region containing the 3243rd mutation in a mitochondrial DNA by a method of using a DNA polymerase.